



ATP2A2 gene

ATPase sarcoplasmic/endoplasmic reticulum Ca2+ transporting 2

Normal Function

The *ATP2A2* gene provides instructions for making an enzyme called sarco(endo)plasmic reticulum calcium-ATPase 2 (SERCA2). This enzyme belongs to a family of ATPase enzymes that helps control the level of positively charged calcium atoms (calcium ions) inside cells. Within the cell, SERCA2 is found in the endoplasmic reticulum and a related structure in muscle cells called the sarcoplasmic reticulum. The endoplasmic reticulum is a structure inside the cell that is involved in protein processing and transport. The sarcoplasmic reticulum assists with muscle contraction and relaxation by releasing and storing calcium ions. Calcium ions act as signals for a large number of activities that are important for the normal development and function of cells. SERCA2 allows calcium ions to pass into and out of the cell in response to cell signals.

Health Conditions Related to Genetic Changes

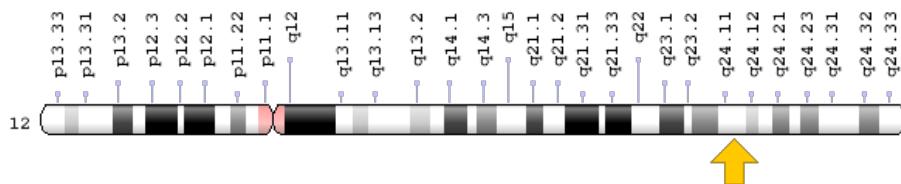
Darier disease

More than 130 mutations in the *ATP2A2* gene have been found to cause Darier disease. Most of these mutations change a single protein building block (amino acid) in the SERCA2 enzyme. All mutations cause the production of a nonfunctional SERCA2 enzyme or cause no SERCA2 to be produced from one copy of the gene. Cells with only one functional copy of the *ATP2A2* gene produce half the normal amount of SERCA2 protein. It is thought that insufficient amounts of SERCA2 combined with outside factors such as heat and minor injury cause the signs and symptoms of Darier disease.

Chromosomal Location

Cytogenetic Location: 12q24.11, which is the long (q) arm of chromosome 12 at position 24.11

Molecular Location: base pairs 110,281,227 to 110,351,093 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AT2A2_HUMAN
- ATP2B
- ATPase, Ca⁺⁺ dependent, slow-twitch, cardiac muscle-2
- ATPase, Ca⁺⁺ transporting, cardiac muscle, slow twitch 2
- calcium-transporting ATPase sarcoplasmic reticulum type, slow twitch skeletal muscle isoform
- sarcoplasmic reticulum Ca(2+)-ATPase 2
- sarcoplasmic/endoplasmic reticulum calcium ATPase 2
- SERCA2
- SR Ca(2+)-ATPase 2

Additional Information & Resources

Educational Resources

- Basic Neurochemistry (sixth edition, 1999): Calcium homeostasis
<https://www.ncbi.nlm.nih.gov/books/NBK27906/figure/A346/>
- Biochemistry (fifth edition, 2002): Structure of SR CA2+ ATPase
<https://www.ncbi.nlm.nih.gov/books/NBK22464/?rendertype=figure&id=A1781>

- Biochemistry (fifth edition, 2002): The Sarcoplasmic Reticulum Ca₂₊ ATPase Is an Integral Membrane Protein
<https://www.ncbi.nlm.nih.gov/books/NBK22464/#A1780>
- Molecular Cell Biology (fourth edition, 2000): Muscle Ca₂₊ ATPase Pumps Ca₂₊ Ions from the Cytosol into the Sarcoplasmic Reticulum
<https://www.ncbi.nlm.nih.gov/books/NBK21481/>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ATP2A2%5BTIAB%5D%29+OR+%28SERCA2%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- ATPase, Ca(2+)-TRANSPORTING, SLOW-TWITCH
<http://omim.org/entry/108740>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_ATP2A2.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=ATP2A2%5Bgene%5D>
- HGNC Gene Family: ATPases Ca₂₊ transporting
<http://www.genenames.org/cgi-bin/genefamilies/set/1209>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=812
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/488>
- UniProt
<http://www.uniprot.org/uniprot/P16615>

Sources for This Summary

- OMIM: ATPase, Ca(2+)-TRANSPORTING, SLOW-TWITCH
<http://omim.org/entry/108740>
- Amichai B, Karpati M, Goldman B, Peleg L. Novel mutations in two families with Darier's disease. Int J Dermatol. 2007 Jan;46(1):64-7.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/17214724>

- Basic Neurochemistry (sixth edition, 1999): ATP-Dependent Ca²⁺ Pumps
<https://www.ncbi.nlm.nih.gov/books/NBK27906/>
- Biochemistry (fifth edition, 2002): Mechanism of P-Type ATPase Action
<https://www.ncbi.nlm.nih.gov/books/NBK22464/?rendertype=figure&id=A1782>
- Dhitavat J, Fairclough RJ, Hovnanian A, Burge SM. Calcium pumps and keratinocytes: lessons from Darier's disease and Hailey-Hailey disease. *Br J Dermatol.* 2004 May;150(5):821-8. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/15149492>
- Miyauchi Y, Daiho T, Yamasaki K, Takahashi H, Ishida-Yamamoto A, Danko S, Suzuki H, Iizuka H. Comprehensive analysis of expression and function of 51 sarco(endo)plasmic reticulum Ca²⁺-ATPase mutants associated with Darier disease. *J Biol Chem.* 2006 Aug 11;281(32):22882-95. Epub 2006 Jun 9.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/16766529>
- Onozuka T, Sawamura D, Yokota K, Shimizu H. Mutational analysis of the ATP2A2 gene in two Darier disease families with intrafamilial variability. *Br J Dermatol.* 2004 Apr;150(4):652-7.
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- Ringpfeil F, Raus A, DiGiovanna JJ, Korge B, Harth W, Mazzanti C, Uitto J, Bale SJ, Richard G. Darier disease--novel mutations in ATP2A2 and genotype-phenotype correlation. *Exp Dermatol.* 2001 Feb;10(1):19-27.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/11168576>

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